

What is Usher syndrome?

Usher syndrome is an inherited disease characterized by partial or total hearing loss and progressive vision loss. Individuals with Usher syndrome have a defect in one of at least nine proteins essential for the development and maintenance of hair cells in the inner ear and light-sensing rod and cone cells in the retina.^{1,2} There are three clinical classes of Usher syndrome, based on the age of onset and severity of symptoms: type I, type II, and type III. The following information focuses on Usher syndrome types IF and IIIA.

What are the symptoms of Usher syndrome, types 1F and IIIA, and what treatment is available?

Symptoms of Usher syndrome type IF begin early in life and may include:³

- Severe to profound hearing loss present at birth or within the first year of life
- Balance problems leading to delays in motor development, sitting, and walking
- Onset of retinitis pigmentosa (progressive vision loss) during childhood with rapid progression to blindness

Symptoms of Usher syndrome type III appear in late childhood or adolescence and are variable. They may include:¹

- Moderate to severe hearing loss starting after speech development and progressing to profound hearing loss by middle age
- Retinitis pigmentosa
- Balance problems

There is no cure for Usher syndrome. Early educational intervention can address sensory and communication needs. While standard hearing aids are usually not helpful, early use of a special hearing aid called a cochlear implant may be considered. A typical lifespan is expected.^{1,4}

How is Usher syndrome, type 1F or IIIA, inherited?

Usher syndrome is an autosomal recessive disease. Usher syndrome type IF is caused by mutations in the *PCDH15* gene, and Usher syndrome type IIIA is caused by mutations in the *CLRN1* gene.^{1,4} An individual who inherits one copy of a *PCDH15* or *CLRN1* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two mutations in the same gene, one from each parent, is expected to be affected. For example, a child with two *PCDH15* mutations is expected to be affected with Usher syndrome type 1F, and a child with one *PCDH15* mutation and one *CLRN1* mutation is a carrier of Usher syndrome type 1F and a carrier of Usher syndrome type IIIA.

If both members of a couple are carriers of a mutation in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Usher syndrome types 1F and IIIA?

Usher syndrome types IF and IIIA can occur in individuals of all races and ethnicities. Both types are known to be more common in individuals of Ashkenazi Jewish ancestry and type IIIA is more common in individuals of Finnish ancestry.¹ Worldwide, Usher syndrome is estimated to affect between 1 in 31,000 to 1 in 16,000 people.³ In the Ashkenazi Jewish population, an estimated 1 in 147 individuals is a carrier of Usher syndrome type IF and 1 in 120 individuals is a carrier of Usher syndrome type IIIA.⁵ In the Finnish population, the carrier frequency of Usher syndrome type IIIA is 1 in 134.⁶

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/usher-syndrome>

National Institute on Deafness and Other Communication Disorders:
<http://www.nidcd.nih.gov/health/hearing/pages/usher.aspx>

Helen Keller National Center for Deaf-Blind Youths and Adults:
<http://www.hknc.org/WhoWeServeUsher.htm>

References

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